Isabelle MARTY
Docteur en Biologie - Ingénieur Chimiste ESPCI

SITUATION PROFESSIONNELLE ACTUELLE

Depuis 2007: Responsable de l'équipe **"Cellular Myology and Pathologies" – Grenoble Institut des Neurosciences" – U 1216**

2009-2011: Directeur adjoint du **"Grenoble Institut des Neurosciences" – U836**

Depuis 2021: Directeur de recherche **INSERM (DR1)**

FORMATION

Avril 2000 : **Habilitation à Diriger les Recherches** (HDR) – Université Grenoble I

1991-1995: **Stage post-doctoral** (Bourse AFM) Laboratoire de Biophysique Moléculaire et Cellulaire. CEA Grenoble

1987-1990 : **Thèse de Biologie** Laboratoire de Biochimie (Pr. P.V. Vignais). CEA Grenoble - "Etude de la topographie membranaire du transporteur mitochondrial d'adénine nucléotides. Approches immunochimique et protéolytique."

1986-1987 : **DEA de Biologie Cellulaire**. Université Joseph Fourier (Grenoble).

1983-1987 : **Ecole Supérieure de Physique et de Chimie Industrielles de la ville de Paris (ESPCI)**. Ingénieur diplômé option chimiste.

COMITES D'EVALUATION

Membre du Conseil Scientifique de l'Association Française contre les Myopathies (AFM) depuis 2012

Expertise scientifique pour l'Inserm, l'Association Française contre les Myopathies (AFM), l'Université Pierre et Marie Curie (Paris), l'Université Lyon I, le "Netherlands Organisation for Scientific Research" (NWO), le Fond National Suisse (FNS), la "RyR1-Foundation"

Reviewer pour American Journal of Physiology, Journal of Biological Chemistry, Journal of Physiology, Journal of Muscle Research and Cell Motility, PLoS One, Neuromuscular Disorders,…

Membre de l'Editorial Board de Frontiers in Skeletal Muscle Physiology depuis 2011

Rapporteur de plus de 20 thèses et HDR, en France et à l'étranger

Membre de la Commission de spécialistes (sections 64-69) de l'Université Grenoble I -1998-2006

Membre de la Commission Recherche des Sciences de la Vie et de la Santé (2000-2003) puis du **Groupe Recherche** de l'UFR de Biologie de l'Université Joseph Fourier (2003-2007)

Expertise externe pour la commission spécialisée Inserm CSS3 (2020; 2023)

DIVERS

Coordinatrice du réseau GIS- Maladies rares (2003-2005) puis **ANR-Maladies rares** (2006-2009) "Bioclinical network for the study of pathologies of the calcium release complex in skeletal muscle"

Organisatrice des X^{èmes} journées Annuelles de la Société Française de Myologie – Grenoble –2012

Membre du comité d'organisation du XIII^{ème} ICNMD (International Congress on Neuromuscular Diseases) - Nice -2014

Membre du comité d'organisation du congrès de la World Muscle Society, Saint Malo, 2017

Organisatrice (chair) de la Gordon Research Conference "Muscle excitation-contraction coupling", Suisse, 2017

Membre du Conseil d'Administration de la Société Française de Myologie –-2012-2021

PUBLICATIONS SELECTIONNEES

Marty I., Robert M., Villaz M., Lai Y., De Jongh K.S., Catterall W.A., and Ronjat M., (1994), *Proc. Natl. Acad. Sci. USA*., **91**, 2270-2274. "Biochemical Evidence for a complex involving Dihydropyridine Receptor and Ryanodine Receptor in Triad Junctions of Skeletal Muscle".

Monnier N, Ferreiro A, **Marty I**, Labarre-Vila A, Mezin P & Lunardi J (2003), *Hum. Mol. Genet.,***12**, 1171-1178 "A homozygous splicing mutation causing a depletion of skeletal muscle RYR1 is associated with MmD congenital myopathy with ophthalmoplegia"

Damy T, Ratajczak P, Shah AM, Camors E, **Marty I**, Hasenfuss G, Marotte F, Samuel J-L, Heymes C, (2004), *Lancet* , **363**, 1365-1367 "Increased neuronal nitric oxide synthase-derived NO production in the failing human heart".

Vassilopoulos S, Thevenon D, …Lacampagne A, Lunardi J, DeWaard M, and **Marty I** (2005), *J. Biol. Chem.*, **280**, 28601-28609. "Triadins are not triad specific proteins: two new skeletal muscle triadins possibly involved in the architecture of sarcoplasmic reticulum."

Oddoux S, Brocard J, …Brocard J, Fauré J, Pernet-Gallay K, Bendahan D, Lunardi J, Csernoch L, **Marty I** (2009) *J Biol Chem*. **284,** 34918-34929 "Triadin deletion induces impaired skeletal muscle function"

Piétri-Rouxel F, Gentil C, Vassilopoulos S,... **Marty I**., Schaeffer L., Voit T. and Garcia L. (2010) *EMBO J*. **29**, 643-54. "DHPR α1S subunit controls skeletal muscle mass and morphogenesis"

Roux-Buisson N., Cacheux M., Fourest-Lieuvin A., Fauconnier J.,…, Probst V., Lacampagne A., Fauré J., Lunardi J., **Marty I.** (2012). *Hum. Mol. Genet*. **21**, 2759-2767. "Absence of Triadin, a Protein of the Calcium Release Complex, is Responsible for Cardiac Arrhythmia with Sudden Death in Human"

Dowling J, Arbogast S…**Marty I**, Lunardi J, Brooks SV, Kuwada JY and Ferreiro A. (2012) *Brain*, **135**,1115-1127. "Oxidative stress and successful antioxidant treatment in models of RYR1-related myopathy"

Dieterich K, Quijrano-Roy S…**Marty I**, Estournet B, Jouk PS, Melki J, Lunardi J. (2013) *Hum Mol Genet* **22,** 1483-1492. "The neuronal endopeptidase ECEL1 is associated with autosomal recessive distal arthrogryposis"

Falcone S, Roman W, Hnia K, Gache V, Didier N, Lainé J, Auradé F, **Marty I** … Gomes ER. (2014) N-WASP is required for Amphiphysin-2/BIN1-dependent nuclear positioning and triad organization in skeletal muscle and is involved in the pathophysiology of centronuclear myopathy. EMBO Mol Med. 6(11):1455-75

Cacheux M, Blum A, Sébastien M, Wozny AS, Brocard J, Mamchaoui K…**Marty I.** (2015) Functional characterization of a Central Core Disease RyR1 mutation (p.Y4864H) associated with quantitative defect in RyR1 protein. *J. Neuromuscular Diseases*, **2**, 421–432

Osseni A, Sébastien M, Sarrault O, Baudet M, Couté Y, …, **Marty I.**, (2016) Triadin and CLIMP-63 form a link between triads and microtubules in muscle cells, *J Cell Sci* **129,** 3744-3755.

Marty I., Fauré J., (2016) Excitation-contraction coupling alterations in myopathies. *J. Neuromuscular Diseases* **3,** 443-453.

Sébastien M, Giannesini B, Aubin P,…, Fauré J, **Marty I**. (2018) Deletion of the microtubule-associated protein 6 (MAP6) results in skeletal muscle dysfunction. *Skelet Muscle*. **8**, 30.

Garibaldi M, Rendu J, Brocard J, Lacene E, Fauré J, …, Taratuto AL, Laporte J, **Marty I**, Antonini G, Romero NB. (2019) 'Dusty core disease' (DuCD): expanding morphological spectrum of RYR1 recessive myopathies. *Acta Neuropathologica Communications* **7**, 3.

Cacheux M, Fauconnier J, Thireau J, Osseni A,… Lacampagne A, **Marty I.** (2019) Interplay between triadin and calsequestrin in the pathogenesis of CPVT in mouse. *Mol Ther*. 2020, 28, 171-179.

Rendu J… **Marty I**, Corne C, Fauré J, Besson G. (2019) Familial deep cavitating state with a glutathione metabolism defect. *Annals of Clinical and Translational Neurology*, 6(12):2573-2578.

Sébastien M, Aubin P., … **Marty I**, Fauré J (2020) Dynamics of the calcium release complex protein triadin within sarcoplasmic reticulum subdomains in skeletal muscle cells *Mol Biol Cell*. **31**:261-272.

Clemens D, Tester D, **Marty I**, Ackerman M. Phenotype-Guided Whole Genome Analysis in a Patient with Genetically Elusive Long QT Syndrome Yields a Novel *TRDN*-Encoded Triadin Pathogenetic Substrate for Triadin Knockout Syndrome and Reveals a Novel Primate-Specific Cardiac *TRDN* Transcript (2020) *Heart Rhythm* **17**, 1017-1024.

Bosson C, Rendu J, Pelletier L, Abriat A, Chatagnon A, Brocard J, Brocard J, Figarella-Branger D, Ducreux S, Van Coppenolle F, Sagui E, **Marty I**, Roux-Buisson N, Faure J, Variations in the *TRPV1* gene are associated to Exertional Heat Stroke. *J Sci Med Sport*. 2020 **23**(11):1021-1027.

Géraud J, Dieterich K, Rendu J, Uro-Coste E, …Madelaine A, **Marty I**, Pégeot H, Thèze C, Siegfried A, Cossée M, Cances C (2020) Clinical phenotype and loss of the slow skeletal muscle troponin T in three new recessive TNNT1 nemaline myopathy patients. *J Med Genet* 2020;**0**:1–7.

Pelletier L, Petiot A, Brocard J, Giannesini B, Giovannini D, Sanchez C, Travard L, Chivet M, Beaufils M, …, Franzini Armstrong C, Romero NB, Rendu J, Jacquemond V, Faure J, **Marty I.** In vivo RyR1 reduction in muscle triggers a core-like myopathy. *Acta Neuropathol. Commun.* 2020 **8,** 192

Beaufils M, Travard L, Rendu J, **Marty I**. Therapies for RYR1-Related Myopathies: Where We Stand and the Perspectives. *Curr Pharm Des*. 2022 **28,**15.

Beaufils M., Tourel A., Petiot A., Halmai N.B., Segal D.J., Rendu J., **Marty I**. Development of knock-out muscle cell lines using lentivirus-mediated CRISPR/Cas 9 gene editing, *J Vis Exp*. 2022 **16**,184.

Wleklinski M, Parikh S, Kryshtal DO, Kim K, **Marty I**, Iyer VR, and Knollmann BC. Impaired dynamic calcium buffering in the sarcoplasmic reticulum causes autosomal dominant CPVT2. *Circ Res*. 2022 **131,** 673.

Chivet M., McCluskey M., Nicot A.S, Brocard J., Beaufils M., Giovannini D., Giannesini B., Poreau B., Brocard J., Humbert S., Saudou F., Fauré J., **Marty I**., Huntingtin regulates calcium fluxes in skeletal muscle *J Gen Physiol*. 2023 **155,** e202213103

Marty I, Beaufils M, Fauré J, Rendu J. Gene therapies for RyR1-related myopathies. *Current Opinion in Pharmacology* in press.